

Counselling Following Diagnosis of a Fetal Abnormality: Comparison of Different Clinical Specialists in Mexico

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Most geneticists agree that counselling should be nondirective, and studies report that genetic counselling by geneticists is performed largely in a neutral style. However, couples at risk of having a child with a genetic condition may seek the advice of other physicians. The purpose of the present study was to describe the answers of four groups of specialists from Mexico City (internists, pediatricians, obstetricians, and neurologists) regarding how they would counsel a couple when prenatal diagnosis has shown that a fetus is affected by one of 17 different genetic disorders and to analyze the role of several variables in the development of their opinion. Our results show that physicians in these specialties are more likely to counsel directly than neutrally. Other variables did not influence the directiveness. With respect to direction of influence, internists, pediatricians, and neurologists are more likely to counsel terminating affected pregnancies than are obstetricians ($P = 0.0002$). Similarly, clinicians older than 37 years of age and those reporting that religion is not important to them counsel terminating affected pregnancies ($P = 0.005$ and $P = 0.003$, respectively). Physicians' gender and clinical experience with genetic diseases did not show statistically significant differences. Strong consensus among specialists was reached only on terminating pregnancies in anencephaly. A lowered and moderate consensus (51–75% agreement) was reached on continuing pregnancies with cleft lip and plate. A moderate measure of consensus for nondirective counselling was found among obstetricians regarding 14 of

the 17 diseases in the study, whereas neurologists expressed a moderate measure of consensus on counselling the termination of pregnancies when the fetus was affected by neurological disorders. Hence, the approach to counselling was related in part to the fetal condition and in part to the clinician's specialty and age and the self-reported importance of religion. The data presented herein may not be representative of all Mexican physicians within the selected specialties; however, it is important to gather their opinions because they are involved in the care and treatment of genetic diseases and may have an important influence on the demand and availability of prenatal diagnosis and abortion. *Am. J. Med. Genet.* 69:23–28, 1997. © 1997 Wiley-Liss, Inc.

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INTRODUCTION

It is generally accepted that genetic counselling is the process of 1) providing the consultant information on a specific genetic disorder, its natural history, complications, possibilities for treatment, mode of inheritance, and recurrence risks and 2) helping couples in their decision-making. In genetic counselling for prenatal diagnosis of genetic disorders and congenital malformations, prospective parents should receive the most complete information so that they are able to provide informed consent for diagnostic procedures. Most geneticists advocate that counselling should be nondirective; that is, it should enable parents to decide for themselves after the best possible counselling [Fraser, 1974; Wertz and Fletcher, 1988]. However, when dealing with prenatal diagnosis, it may be difficult to maintain a neutral position if a fetal abnormality is found [Clarke, 1991]. Thus, when prenatal diagnosis detects a fetal abnormality, parents are faced with the particularly

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difficult decision between continuing or terminating pregnancy. The hardest process involves weighing severity and treatability of particular genetic disorders in the face of uncertain prognosis [Fletcher and Wertz, 1992]. The parents' perception of the severity of the disorder and their decision to abort may be influenced by the counselling style, by what they are told by the counselors regarding the condition [Robinson et al., 1989], and by the medical specialty of the counselor [Verp et al., 1988].

The extent to which physicians and health professionals dealing with genetic disorders are nondirective in their counselling approach has been studied previously, showing differences related to profession, medical specialty, gender, religion, culture, and nation [Wertz et al., 1990; Renaud et al. 1993; Marteau et al., 1994a,b]. In Mexico, as in other countries, geneticists have generally been trained to provide nondirective genetic counselling; however, they have little involvement in the medical care of the patients. Geneticists participate in the diagnosis of genetic disorders, provide counselling to the family on as many occasions as needed, and refer the patients to other specialists for specific medical and surgical treatments. Therefore, nongeneticists, who are typically directive when recommending the best treatment, are the ones who share with the patients and their families the problems and consequences of the disorder as well as the anguish of long and costly treatments. As a consequence, they may become involved in prenatal diagnosis and in decisions concerning terminating or continuing the pregnancy after a diagnosis of fetal abnormality. The demand for genetic services and prenatal diagnosis is growing; however, Mexican law has limited the performance of the latter because terminating a pregnancy on the basis of a fetal defect or disease is illegal in most states of the country. Only one public and a few private Mexican institutions have been performing prenatal diagnosis, and the procedure is offered as an option to a very small number of families with a high risk of genetic disorders. Mexican physicians are beginning to face ethical problems in this area, and very little is known about their attitudes and positions regarding many aspects of genetic counselling, prenatal diagnosis, and selective abortion. The purpose of the present study is to describe the answers of four groups of specialists with respect to how they would counsel a couple when the prenatal diagnosis has shown that the fetus is affected by a genetic disorder and to analyze the role of several variables in determining their opinion. The four groups see, with variable frequency, patients of different ages with genetic disorders.

Our study design is based on the studies published by Marteau et al. [1994a,b], who used a questionnaire to explore the approach to counselling women following the diagnosis of a fetal abnormality by three groups of health professionals (genetic nurses, geneticists, and obstetricians) and by geneticists of different countries (Germany, Portugal, and the United Kingdom). We were interested in comparing how four different types of clinical specialists from our country would answer a verbatim translation of the same questionnaire.

MATERIALS AND METHODS

Samples

The following clinical specialists were chosen from four of the National Institutes of Health in Mexico City, which are public health institutions providing mainly tertiary medical care and which are teaching and research centers. 1) The first group was internists; a randomized sample of 33% (60 physicians) was selected from among the internal medicine specialists employed at the Instituto Nacional de la Nutrición, where only adult patients are seen and very few of these have genetic diseases; 56 agreed to participate. 2) The second group was pediatricians; a random sample of 33% (59 physicians) was selected at the Instituto Nacional de Pediatría; these doctors frequently attend patients with genetic diseases or congenital malformations [Carnevale et al., 1985]; 58 agreed to participate. 3) The third group was gynecologists and obstetricians; all of these specialists (47 physicians) at the Instituto Nacional de Perinatología participated in the study; this is the only public institution in Mexico where a prenatal diagnosis program is conducted. 4) The final group was neurologists; the sample included 26 of the 30 neurologists at the Instituto Nacional de Neurología y Neurocirugía, where adult patients with neurological disorders are seen.

Because the authors of this report work in the above-mentioned Institutes and the invitation to participate and the questionnaire were personally distributed, the participants were asked to fill out the questionnaire anonymously. If the questionnaire was not returned within 15 days, a reminder telephone call was made. The number and general characteristics of the respondents at each Institution are shown in Table I.

Procedures

The questionnaire was a Spanish translation of the multiple-choice questionnaire of Marteau et al. [1994a,b]. Physicians were asked to state how they would counsel a couple found to carry a fetus with one of 17 diseases (the list is shown in Table IV). All questions had the same five options as answers: 1) encourage parents to carry the pregnancy to term; 2) try to be as neutral as possible, but stress the more positive than the negative aspects of the disease; 3) try to be as neutral as possible, covering both positive and negative aspects; 4) try to be as neutral as possible, but stress the more negative than the positive aspects of the disease; and 5) encourage termination of pregnancy.

The extent of directiveness in the approach to counselling and the direction towards continuing or terminating the affected pregnancy were assessed. The scale of directiveness used was as reported by Marteau et al. [1994a,b]. In short, a value of 2 was assigned to option 3 and a value of 1 for options 2 and 4. The extreme options, either 1 or 5, were coded as 0. The theoretical limits of this scale ranged from 0 (counselling continuation or termination of the pregnancy for the 17 conditions) to 34 (nondirective counselling for all conditions).

The direction of counselling was established by scoring the answers in the following manner: 1 = +2, 2 = +1, 3 = 0, 4 = -1, 5 = -2. A mean score above 0 for

TABLE I. Characteristics (%) of Respondents

Characteristic	Specialty				χ^2	<i>P</i>
	Internists (n = 56)	Pediatricians (n = 58)	Obstetricians (n = 47)	Neurologists (n = 26)		
Sex						
Male	86	62	87	88	14.9	0.001
Female	14	38	13	12		
Age						
≤ 37 years	41	53	54	38	3.4	0.328
> 37 years	59	47	46	62		
Importance of religion						
Important	61	74	81	81	6.7	0.081
Not important	39	26	19	19		
Attending genetic disease						
Frequently	11	57	17	38	34.6	<0.001
Rarely	89	43	83	62		

each respondent across all 17 conditions indicates a tendency to counsel toward continuing affected pregnancies, whereas a score below 0 indicates a tendency to counsel towards terminating affected pregnancies. A score of 0 can be obtained if counselling is nondirective or if an equal number of respondents counsel in both directions.

Statistical Analysis

Statistical analysis included estimation of differences among medical specialties and pairwise comparison of other variables, such as gender, age, importance of religion, and frequency of contact with patients with genetic disorders; the χ^2 statistic was used. Comparison of directiveness and direction of directiveness were evaluated by the Kruskal-Wallis one-way analysis of variance, which is a nonparametric test that is very useful in deciding whether the differences seen in more than two independent samples indicate true differences among populations or simple random variations.

In an additional analysis, we recoded the direction of directiveness in two categories: 1 = the sum of -1 and -2 and 0 = the sum of $+1$ and $+2$ (the nondirective responses were excluded). Thus, by means of unconditional logistic regression analysis (multivariate), we estimated the probability of making negative decisions, compared to positive decisions, according to specialty, gender, age, importance of religion, and frequency with which the physicians see patients with genetic disorders. The association measure reported is the odds ratio (OR) derived from the exponential of regression coefficients.

Consensus within a specialty and among specialties was assessed. A strong consensus was defined on the basis of the “ $\frac{3}{4}$ rule” of Wertz [1990], which requires an agreement among more than 75% of respondents; agreement between 51% and 75% of respondents was considered to be moderate consensus.

RESULTS

Comparison of the general characteristics of respondents according to their specialties showed statistical differences in the male-female ratio and in the frequency with which the respondents attend patients

with genetic disorders. This is because pediatricians had a higher proportion of women and saw more patients with genetic disorders than other specialists. On the other hand, specialists were similar with respect to age and the importance of religion in their everyday life (Table I).

Directiveness and Direction of Directiveness

Directiveness measured the extent to which counselling was directive, and all four groups were directive. No significant differences in directiveness were found with regard to medical specialty, gender, age, and frequency with which patients with genetic disorders are seen. Only religion showed a borderline significance (Table II), with a lower score in respondents who did not consider religion as important.

With respect to the direction of counselling, obstetricians were more likely to counsel towards continuing affected pregnancies than were the other specialists ($P = 0.0002$; Table II). On the other hand, clinicians older than 37 years of age and those reporting that religion was not important showed a tendency to counsel towards terminating affected pregnancies ($P = 0.005$ and $P = 0.003$, respectively; Table II). Gender and frequency of contact with patients with genetic disorders did not result in statistically significant differences.

Probability of Counselling Termination of Affected Pregnancies

In the multivariate logistic regression analysis (Table III), we found that three variables, specialty, age, and religious views, selected in a stepwise model, were associated with the probability of counselling towards termination of affected pregnancies. Internists, pediatricians, and neurologists exhibited a higher probability of counselling towards termination than did obstetricians (OR = 2.13, 1.88, and 5.64, respectively). Age over 37 years was associated with a higher probability than age under 37 years (OR = 2.05), and importance of religion was associated with a lower probability (OR = 0.34).

Consensus

As is shown in Table IV, a strong consensus among specialists was evident only for encouraging termination

TABLE II. Mean Scores for Directiveness and Direction of Directiveness in Counselling

	Directiveness		Direction	
	Mean (S.D.)	<i>P</i> value ^a	Mean (S.D.)	<i>P</i> value ^a
Specialty		0.34		0.0002
Internists	18.8 (8.0)		-0.12 (0.73)	
Pediatricians	19.1 (7.7)		-0.09 (0.71)	
Obstetricians	21.0 (7.8)		0.20 (0.53)	
Neurologists	18.3 (9.7)		-0.50 (0.63)	
Gender		0.33		0.18
Male	19.0 (8.5)		-0.12 (0.72)	
Female	20.7 (6.4)		0.04 (0.59)	
Age		0.27		0.005
≤37 years	19.7 (8.4)		0.06 (0.71)	
>37 years	19.1 (7.9)		-0.21 (0.66)	
Importance of religion		0.07		0.003
Important	19.8 (8.5)		0.02 (0.70)	
Not important	18.2 (7.2)		-0.34 (0.61)	
Attending genetic disease		0.57		0.61
Frequently	18.7 (8.1)		-0.03 (0.78)	
Rarely	19.7 (8.1)		-0.10 (0.65)	

^a Obtained by Kruskal-Wallis one-way ANOVA.

of pregnancy when the fetus was anencephalic. A moderate consensus was found for encouraging continuation of the pregnancy for cleft lip and palate. When examining consensus within the specialties, strong consensus (95.2%) to encourage continuing pregnancy was evident for cleft lip and palate among obstetricians. Also, in the four specialties, almost 90% of the respondents were likely to encourage termination of pregnancy for anencephaly.

Moderate consensus on nondirective counselling was observed among obstetricians, more than 50% of whom were neutral on the subject of 13 of the 17 diseases in the survey, whereas internists, pediatricians, and neurologists reached moderate consensus for only two, one and three conditions, respectively (Table IV). Neurologists showed moderate consensus on counselling towards terminating affected pregnancies for five conditions: Huntington disease, 50% risk of Alzheimer disease, Alzheimer disease, achondroplasia, and Duchenne muscular dystrophy.

DISCUSSION

Our results show that internists, pediatricians, obstetricians, and neurologists participating in the study

TABLE III. Multivariate Model Obtained by Logistic Regression (Stepwise Method) to Estimate the Probability to Make Negative Decisions (-1, -2) Compared to Positive Decisions (+1, +2), Excluding Nondirective Decisions

Variable	OR	95% CI	<i>P</i> value*
Specialty			
Internists vs. obstetricians	2.13	1.02-4.48	0.05
Pediatricians vs. obstetricians	1.88	0.91-3.87	0.09
Neurologists vs. obstetricians	5.64	1.78-17.9	0.003
Age (years):			
>37 vs. ≤37	2.05	1.09-3.87	0.03
Importance of religion:			
Important vs. nonimportant	0.34	0.19-0.62	<0.001

*Model *P* value = 0.008.

were more likely to counsel directly than neutrally. The tendency to be neutral was not influenced by gender, age, or the frequency with which these clinicians see genetic diseases. Clinicians were more likely to be neutral when religion was important to them than when religion was unimportant.

In comparison to the results of Marteau et al. [1994b], it is clear that British geneticists and genetic nurses (mean scores 24.9 and 28.4, respectively) were much more neutral than our four groups of specialists, whereas British obstetricians had a similar mean score of directiveness (17.0). On the other hand, geneticists from three European countries reported that they approach counselling in a broadly nondirective style: Mean scores and SDs were 26.2 ± 6.6 for the United Kingdom, 21.2 ± 6.4 for Portugal, and 21.1 ± 8.0 for Germany [Marteau et al., 1994a]. The same conclusion regarding nondirective counselling by geneticists was obtained in a survey of 18 countries [Wertz et al., 1990]. Clinical training may explain these differences, because geneticists are generally trained to provide neutral genetic counselling based on respect for patient autonomy, whereas other specialists are trained to make decisions on patient treatment. Also, there are significant cross-cultural differences in the practice of prenatal diagnosis for fetal anomalies. For example, ethnically English physicians from the province of Quebec, Canada, involved in prenatal diagnosis (obstetricians, pediatricians, and radiologists) were more likely to be neutral than ethnically French physicians from the same province and in comparison to French physicians [Renaud et al., 1993]. In this survey, the authors found that, the more religious the physician is and the older he or she is, the more he or she will tend to be directive.

For directive counselling, our results showed that specialty was related to the direction of the counselling. Internists, pediatricians, and neurologists were more likely to counsel towards terminating affected pregnancies than did obstetricians, who reported counselling towards continuing affected pregnancies. In

TABLE IV. Percentage of Specialists Reporting Counselling Following the Prenatal Diagnosis of 17 Different Conditions

Condition	Tendency											
	Neutral ^a				Continuing affected pregnancy ^b				Terminating affected pregnancy ^c			
	1 ^d	2	3	4	1	2	3	4	1	2	3	4
Cleft lip-palate	30	29	4	19	68	67	96	69	2	4	0	11
Open spina bifida	36	48	62	38	16	17	19	11	48	35	19	50
Closed spina bifida	37	38	40	38	38	53	56	35	25	9	4	27
Anencephaly	11	2	2	8	3	3	7	4	86	95	91	88
Cystic fibrosis	35	50	50	44	21	15	23	8	44	35	27	48
Sickle cell anemia	50	52	58	50	39	21	36	15	11	27	6	35
Huntington disease	32	45	64	31	19	14	19	4	49	41	17	65
50% Alzheimer disease	40	48	55	31	28	24	30	15	32	28	15	54
Alzheimer disease	29	49	61	38	25	16	22	8	46	35	17	54
Turner syndrome	54	44	52	35	16	26	33	15	30	30	15	50
Down syndrome	41	37	55	42	14	25	21	12	45	38	24	46
Klinefelter syndrome	53	48	54	42	21	17	27	12	26	35	19	46
Achondroplasia	37	48	56	38	20	19	23	0	43	33	21	62
PKU	44	43	57	54	28	28	34	11	28	29	9	35
Hemophilia	41	46	62	54	27	26	27	11	32	28	11	35
PKD	47	44	64	54	22	17	26	4	31	39	10	41
DMD	46	40	52	31	8	12	13	4	46	48	35	65

^a Percentage of respondents choosing response option 3.^b Percentage of respondents choosing response options 1 or 2.^c Percentage of respondents choosing response options 4 or 5.^d 1, internists (n = 56); 2, pediatricians (n = 58); 3, obstetricians (n = 47); 4, neurologists (n = 26).

contrast, British obstetricians were significantly more likely to counsel towards terminating affected pregnancies (mean score -0.32) than were geneticists and genetic nurses [Marteau et al., 1994b]. Here, the interesting difference is between British and Mexican obstetricians. In our study, the tendency of obstetricians to counsel toward continuing affected pregnancies may be due to the fact that obstetricians are more directly involved than other clinicians—the obstetrician performs the procedure—if the couple decides to terminate the pregnancy. The lack of legal protection for performing abortion in cases of fetal abnormality in most states of the country, including Mexico D.F., where the survey was done, may partially account for the obstetricians' reluctance to counsel for termination. This statement is consistent with the results of Geller et al. [1993] from France. French law legalizes selective termination, setting no upper gestational age limit on the abortion of fetuses with anomalies, and the majority of Parisian obstetricians would approve second- and third-trimester termination of fetuses with spina bifida, trisomy 21, microcephaly, and Duchenne muscular dystrophy. This attitude toward termination was associated with their reported practice of recommending abortion of abnormal fetuses.

Marteau et al. [1994a] found that Portuguese geneticists were more likely to counsel towards terminating pregnancies than were their counterparts in the United Kingdom and Germany; the authors argue that economic factors may shape counselling in different countries. Our results show that our specialists, except for obstetricians, reported a tendency to counsel towards termination of pregnancies. It may be that severe social, educational, and economic problems of Mexican families with genetic disorders and congenital malformations

attending the public hospitals where the survey was performed influence clinicians to act in favor of preventing suffering and not placing a burden on these families, as opposed to remaining neutral and leaving the decision to the couple. The lack of availability of health care and educational programs for the disabled in our country is another factor that may influence this paternalistic attitude.

A surprising finding, for which we have no ready explanation, was that specialists older than 37 years of age were more likely to counsel towards terminating affected pregnancies than those under age 37 years. However, it is possible that new treatments or new perceptions of disability led to greater optimism among younger specialists. On the other hand, as expected, religion had an important effect on the tendency of counselling. When specialists are more religious, the probability of counselling towards termination of pregnancy is significantly lower than when they are not religious. In Mexico, most people are Catholic, and the Catholic church is known to be strongly opposed to abortion under any circumstances.

A great variability in responses was found, and strong consensus among clinicians was reached only for anencephaly, for which more than 80% of physicians would give pessimistic information or encourage termination of pregnancy. A lowered consensus, from strong to moderate, was reached for pregnancies that continued with fetuses with cleft lip and palate, a condition generally perceived as mild. Obstetricians reached moderate consensus on counselling nondirectively in 13 of 17 conditions in the survey, indicating a tendency to be more neutral than the other specialists. On the other hand, neurologists reached moderate consensus on counselling towards terminating affected

pregnancies for achondroplasia and for four degenerative neurological disorders that are commonly seen by them. The close relationship with patients, affected by these disorders, possibly influenced the neurologists' perceived severity of the diseases.

The data presented herein are biased by several limitations of the methodology. 1) The samples, at best, may be representative of the specialists working in the selected institutions, and not of all the specialists in the country, because these medical centers are characterized by access to modern technology, provision of mainly tertiary medical care, teaching, and research. 2) Patients attending these public institutions have, in general, low incomes and low educational levels and may accede to the physicians' highly prestigious position and paternalistic attitudes, making directiveness more acceptable. 3) Another important note of caution concerns the fact that, except for obstetricians who have clinical experience with prenatal diagnosis, clinicians have very little experience in such situations, and their responses were based mostly on theoretical grounds. 4) We did not assess the knowledge of the respondents with regard to the disorders in the questionnaire, and this would influence the thinking about what to counsel in a specific situation.

In spite of these limitations, it is important to explore the counselling tendencies of clinicians and the role of several variables as determinants of their opinions, because the number of geneticists is still very small and the opinion of other specialists involved in care and treatment of patients with genetic disorders may have an important impact. These clinicians may influence positively or negatively the decisions of patients, as has been shown in other studies [Holmes-Siedl et al., 1987; Robinson et al., 1989]; the demand for prenatal diagnosis and abortion; the availability of these services; and, also, the possibility of legalization of abortion on the basis of a fetal defect.

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